Antenatal and Newborn Screening

NHS
Antenatal and Newborn Screening Programmes

The UK National Screening Committee (NSC)

Annie Roberts

NSC
UK National Screening Committee

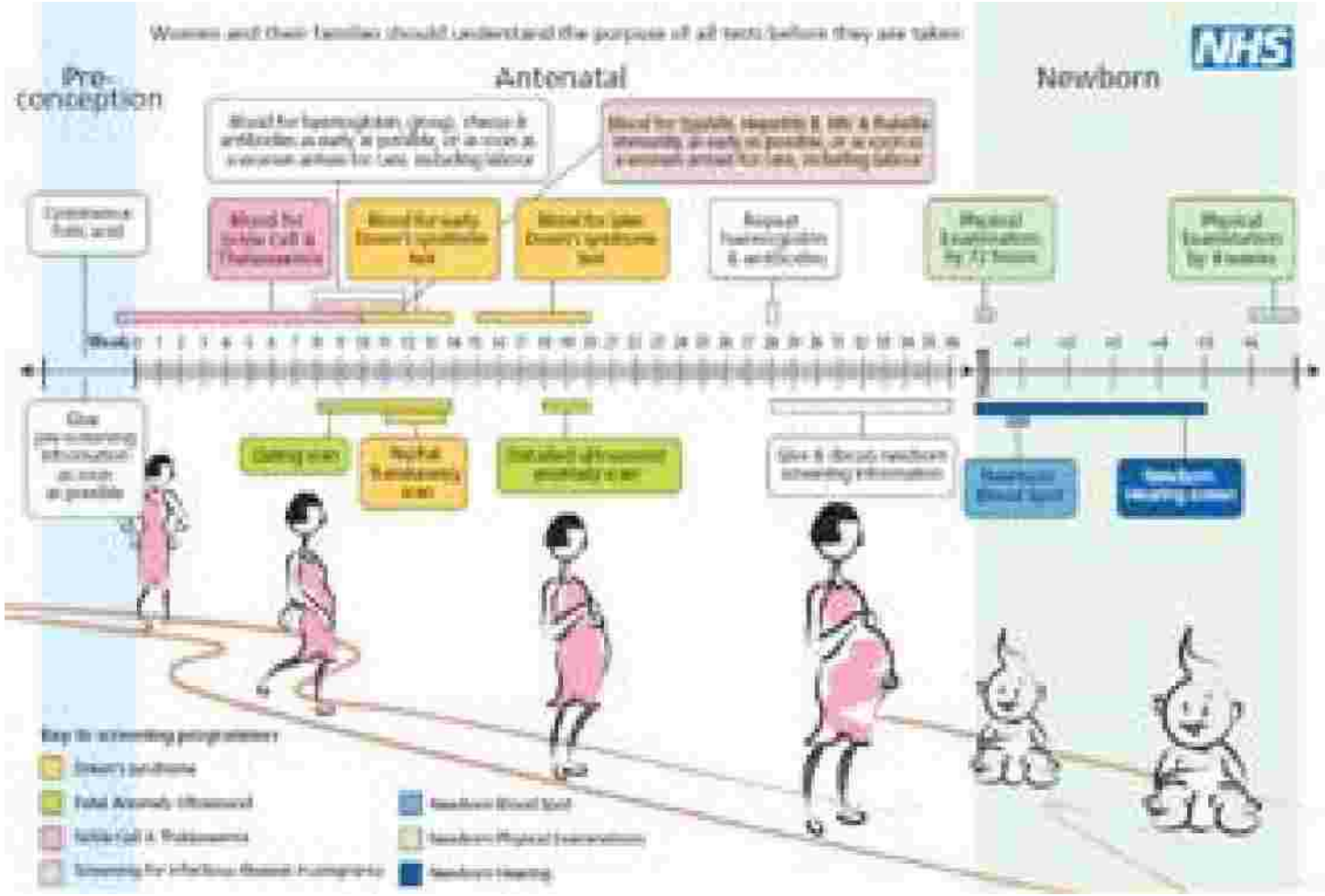
Contact details

Antenatal and Newborn Screening Programmes

- Level 5 Women's Centre JR
- Tel 01865 221087
- Mobile 07909988993
- Email anne.roberts@orh.nhs.uk

UK National Screening Committee

Screening Timeline

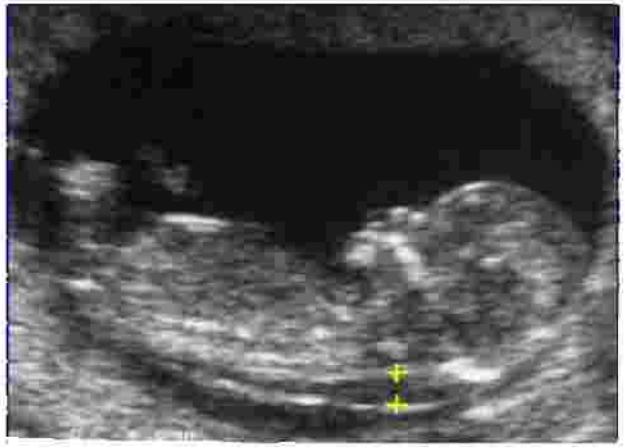


Patient information

NHS
Antenatal and Newborn Screening Programmes

NSC
UK National Screening Committee

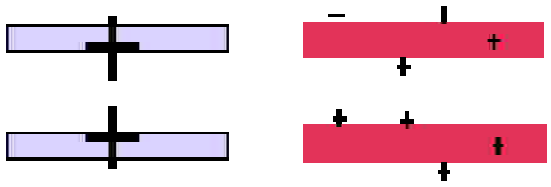
Nuchal Translucency



- Measured between 11 - 13+6 weeks

Positioning of callipers

- Very precise measurements needed
- Position of fetus and Maternal habitus are significant factors in obtaining this measurement



First Trimester Serum Markers

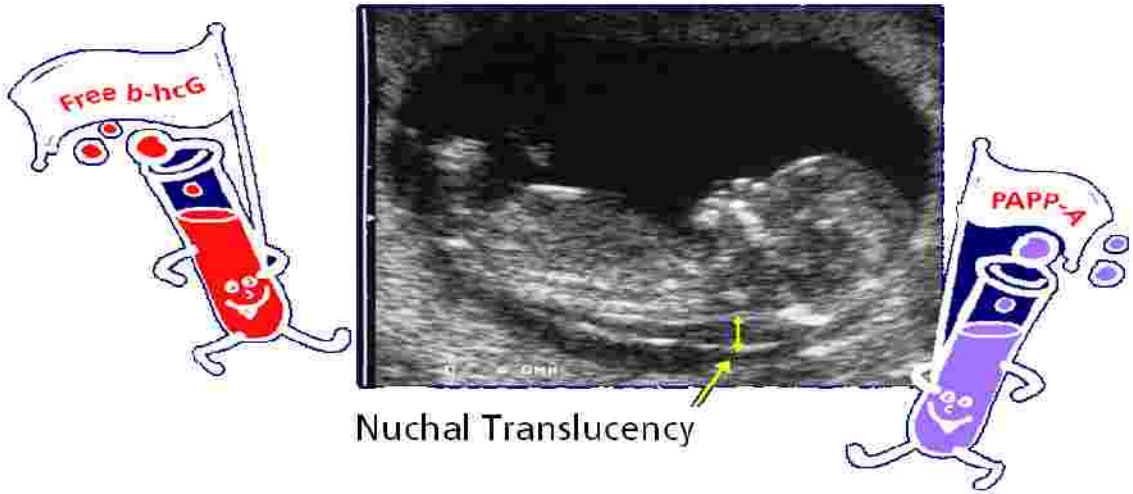
- PAPP-A
  - **P**regnancy **A**ssociated **P**lasma **P**rotein-**A**
- Originates mainly from placenta syncytiotrophoblasts
- Concentration increases with gestation
- Screening sensitivity decreases with gestation
- Optimal sensitivity 10-11 weeks gestation
- Levels reduced (0.34-0.58 MoM) in affected pregnancies

First Trimester Serum Markers

- bhCG
  - b subunit of **h**uman **C**horionic **G**onadatrophin
- Produced by syncytiotrophoblast cells
- Concentration decreases with gestation
- Sensitivity maintained with gestation
- Levels increased (2.2 MoM) in affected pregnancies

Combined Test

Timing 11-13+6 weeks

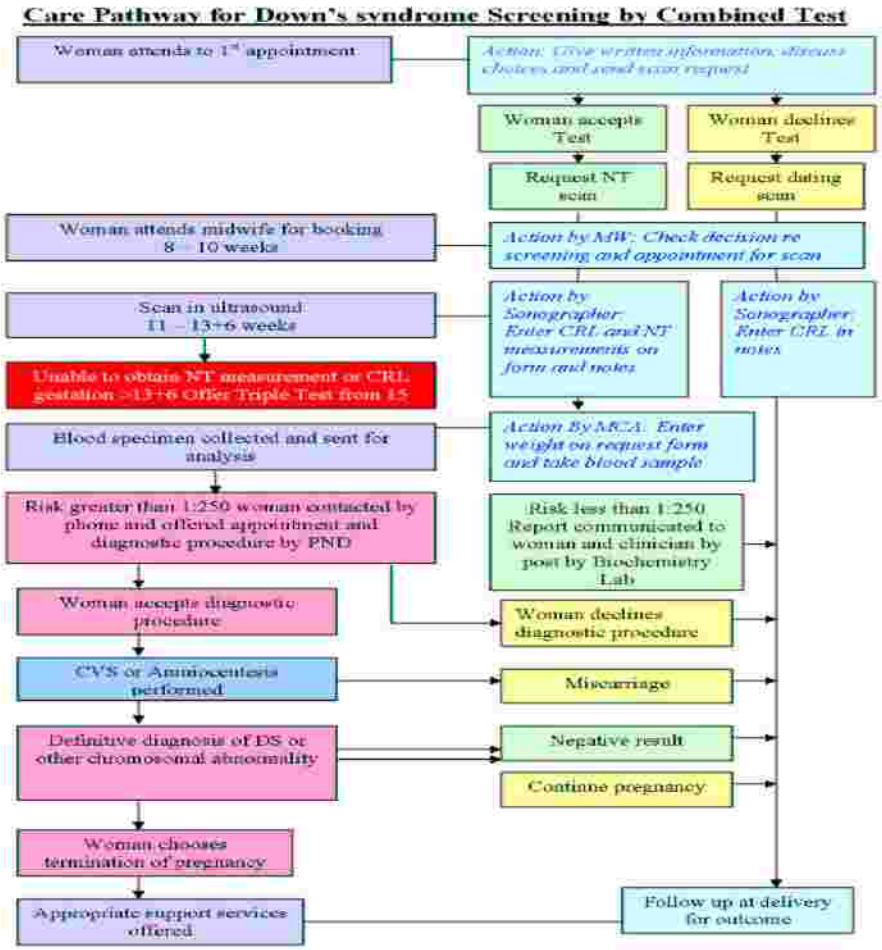


Factors affecting screening for Down's syndrome?

- Maternal weight
- Ethnic group
- Prev pregnancy history
- Smoking
- Insulin dependant diabetes
- Multiple pregnancy
- Assisted conception
- Bleeding in pregnancy

Care Pathway for Combined Screening

NHS
Antenatal and Newborn Screening Programmes



NSC
UK National Screening Committee

What if NT cannot be obtained (too early)

- If CRL measurement gives gestation <11 weeks
- Pregnancy dated and appointment booked for correct gestation

What if NT cannot be obtained

- If CRL measurement gives gestation >13+6 weeks
- If specific image unobtainable due maternal habitus, retroverted uterus or fetal position
- Pregnancy dated, information entered on request card and date suggested for Triple Test

- Triple Test will be available for women who book late or NT cannot be obtained
- Taken at 15-19+6 weeks
  - Dating scan
- Current performance of Triple Test
  - Detection rate 100%
  - FPR 5%

AFP only

- Requests for this test to screen for neural tube defects will no longer be accepted in line with National guidelines
- The exception will be requests made by PND for previous history.
- It will still form part of the Triple Test so an occasional raised AFP may be reported

Infectious Diseases Screening

NHS Antenatal and Newborn Screening Programmes

Hepatitis B

- Virus carried in blood
- Passed by blood-to-blood contact (eg sex, needles)
- Reduce vertical transmission
- Baby immunised
- Can be asymptomatic

Syphilis

- Sexually transmitted
- Cured with antibiotics
- Outcomes improved if detected
- Can be asymptomatic

HIV

- Virus that can cause AIDS
- Passed by blood-to blood contact (eg sex, needles)
- Reduce vertical transmission
- Can be asymptomatic

Rubella

- Check immunity
- Outcomes improved through testing
- Postnatal vaccination for future protection

- HIV
  - Uptake consistently above 98% target
  - 96-98% from 59% in 2004
  - 12-18 women booked per year are HIV positive
  - Approximately 50% new diagnosis
  - No cases of vertical transmission recorded in screening women

- Hepatitis B
  - >99% uptake
  - 10-15 women booked per year are positive
  - Approximately 60% new diagnosis
  - Improved compliance with immunisation programme for infants >95% have completed the full course and gaining immunity

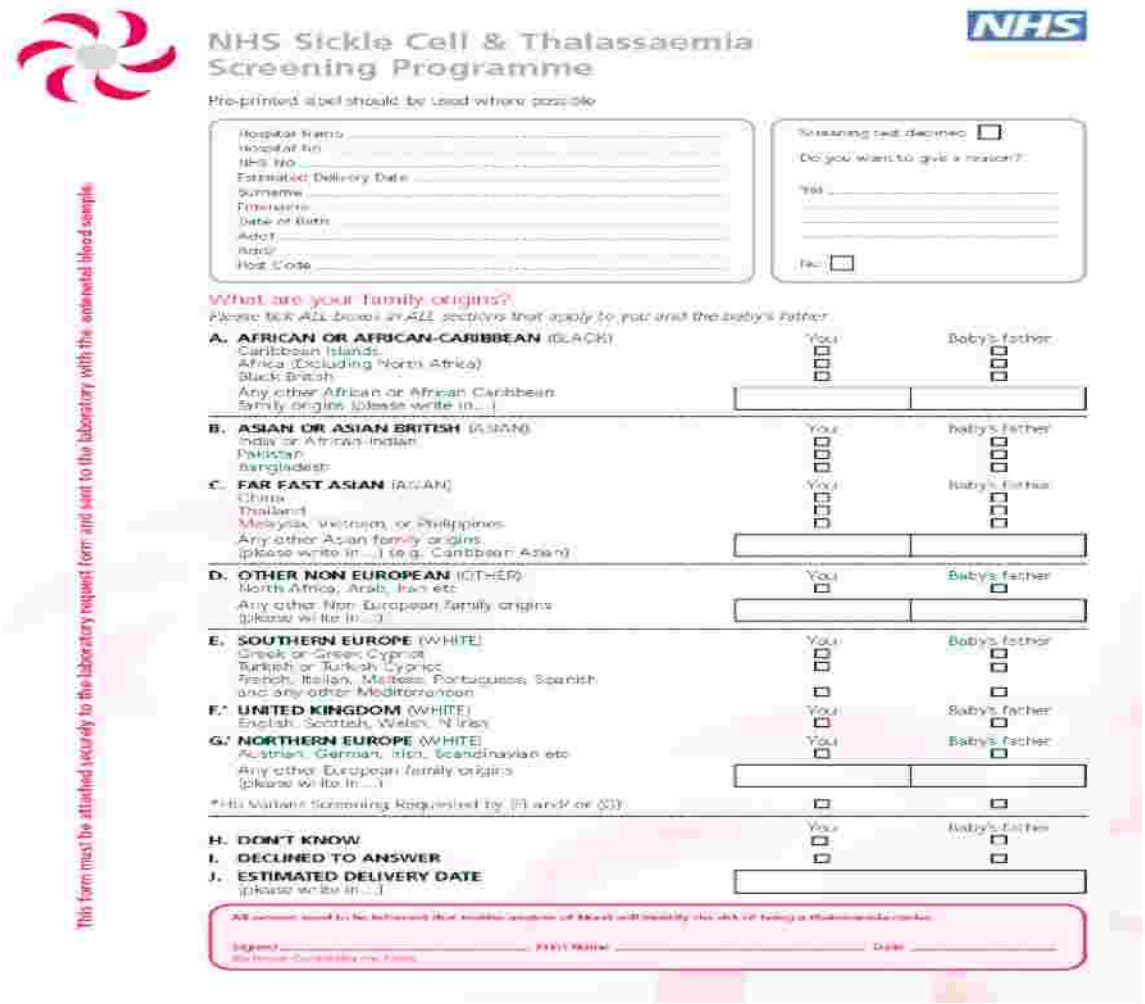
- Syphilis
  - >99% uptake
  - 8-10 women booked per year are positive
  - 1 case previously untreated this year, others are old infections

- Rubella
  - >99% uptake
  - Non immune women numbers rising approx 10%
  - Offer MMR post delivery
  - Immunisation of non immune women to be audited to assess uptake

- Screen offered on FBC sample at booking
- Activate by completing Family Origins Questionnaire and enclosing with sample
- If partner screening required will be activated via screening coordinator
- High risk couple will be referred to PND
- Linked with Newborn programme

Sickle Cell Disease and Thalassaemia Screening

Antenatal and Newborn Screening Programmes



NHS Sickle Cell & Thalassaemia Screening Programme

Pre-printed label should be used where possible

Hospital Name
Hospital No
NHS No
Estimated Delivery Date
Surname
Forename
Date of Birth
Add1
Add2
Post Code

Screening test declined ☐
Do you want to give a reason?
Yes
No ☐

This form must be attached securely to the laboratory request form and sent to the laboratory with the antenatal blood sample

What are your family origins?

Please tick ALL boxes in ALL sections that apply to you and the baby's father

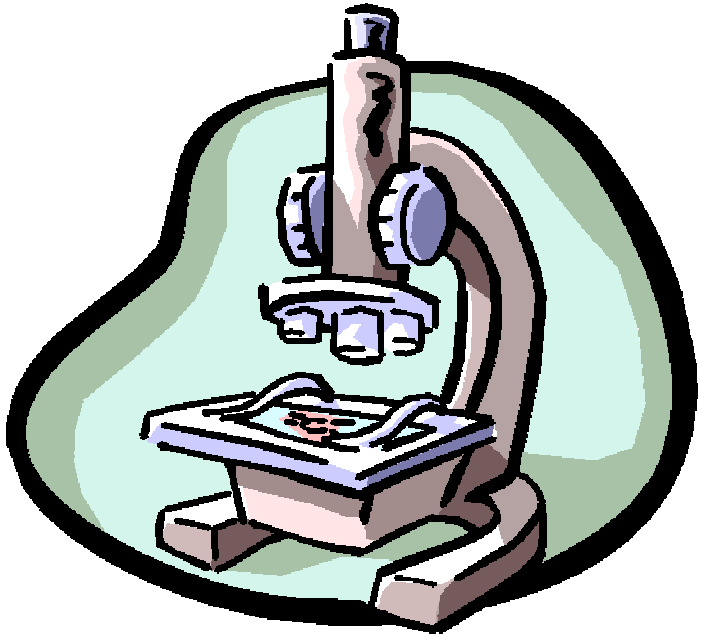
	You	Baby's father
A. AFRICAN OR AFRICAN-CARIBBEAN (BLACK)		
Caribbean Islands	☐	☐
Africa (Excluding North Africa)	☐	☐
Black British	☐	☐
Any other African or African Caribbean family origins (please write in....)		
B. ASIAN OR ASIAN BRITISH (ASIAN)	You	Baby's father
India or African-Indian	☐	☐
Pakistan	☐	☐
Bangladesh	☐	☐
C. FAR EAST ASIAN (ASIAN)	You	Baby's father
China	☐	☐
Thailand	☐	☐
Malaysia, Vietnam, or Philippines	☐	☐
Any other Asian family origins (please write in....) (e.g. Caribbean Asian)		
D. OTHER NON EUROPEAN (OTHER)	You	Baby's father
North Africa, Arab, Iran etc	☐	☐
Any other Non European family origins (please write in....)		
E. SOUTHERN EUROPE (WHITE)	You	Baby's father
Greek or Greek Cypriot	☐	☐
Turkish or Turkish Cypriot	☐	☐
French, Italian, Maltese, Portuguese, Spanish and any other Mediterranean	☐	☐
F. UNITED KINGDOM (WHITE)	You	Baby's father
English, Scottish, Welsh, N Irish	☐	☐
G. NORTHERN EUROPE (WHITE)	You	Baby's father
Austrian, German, Irish, Scandinavian etc	☐	☐
Any other European family origins (please write in....)		
*Hb Variant Screening Requested by (F) and/or (G)	☐	☐
	You	Baby's father
H. DON'T KNOW	☐	☐
I. DECLINED TO ANSWER	☐	☐
J. ESTIMATED DELIVERY DATE (please write in....)		

All women need to be informed that routine analysis of blood will identify the risk of being a thalassaemia carrier

Signed _____ Print Name _____ Date _____
(by Person Completing this Form)

NSC UK National Screening Committee

Haemoglobin Percentages in Adults



- Hb A : 95 - 98%
- Hb F: 1% (approx.)
  - Can rise to 5% in pregnancy without concern)
- Hb A_2 : 1.8 - 3.4%

Antenatal Screening

Significant Maternal Hb'opathies

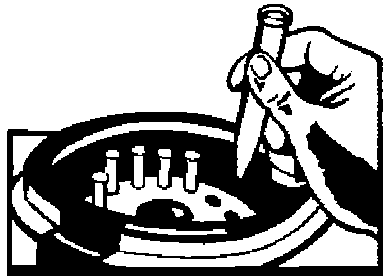
- SCD
- b Thalassaemia intermedia
- Hb H disease
- b Thalassaemia major (already apparent)

Significant Carrier States

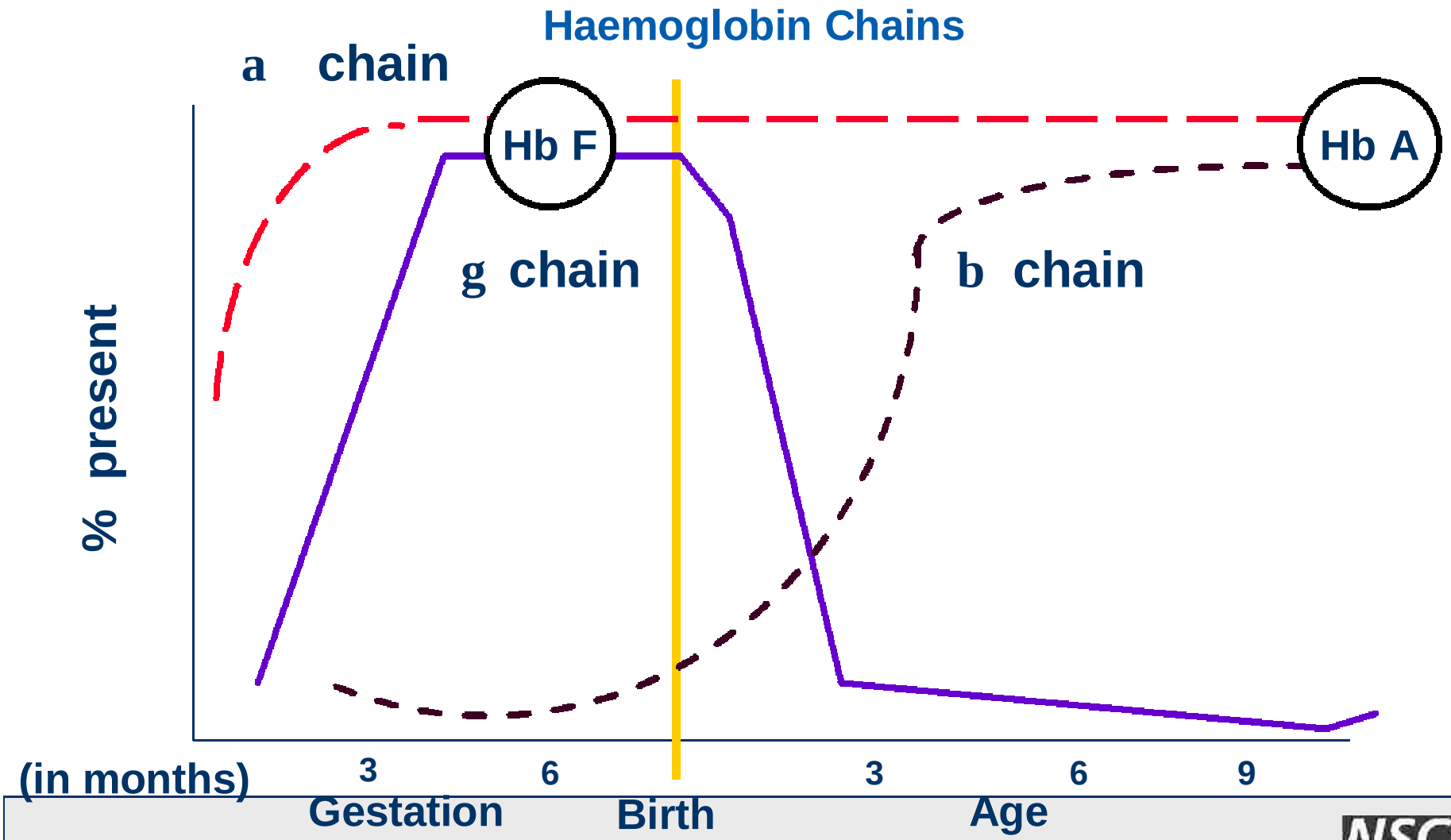
- Hb AS
- Hb AC
- Hb AD
- Hb AE
- Hb AO Arab
- Hb A Lepore
- b Thalassaemia trait
- db Thalassaemia trait
- a^0 Thalassaemia trait
- HPFH

Other Significant Conditions

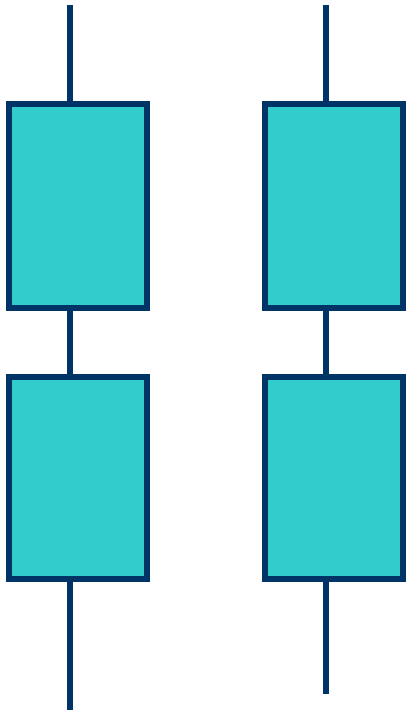
- Compound heterozygote conditions
- Homozygous conditions



All require partner testing

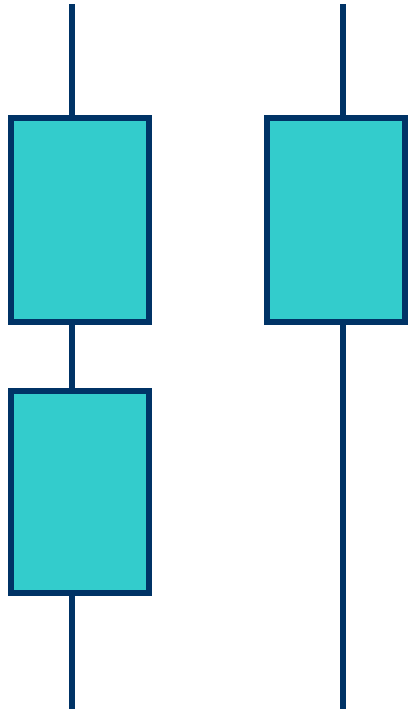

Haemoglobin Chains
a chain
Hb F
Hb A
g chain
b chain
% present
(in months)
3
6
3
6
9
Gestation
Birth
Age

Alpha Thalassaemia



4 a globin genes

a a / a a

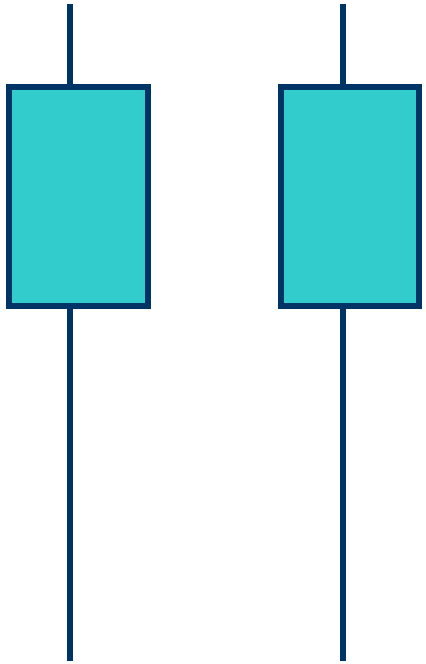


Heterozygous a $^{+}$ Thalassaemia (Carrier)

a a / a -

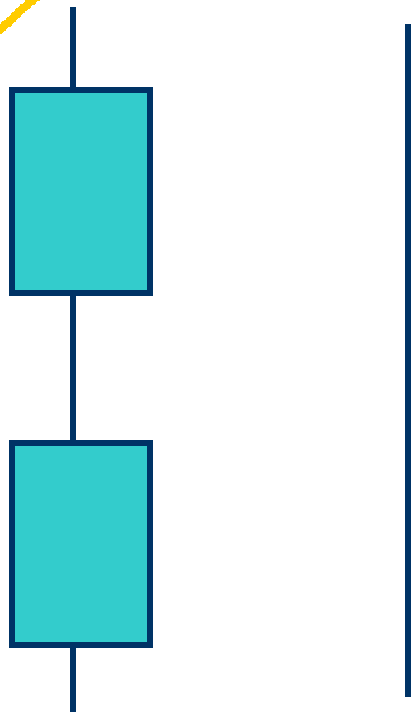
Alpha Thalassaemia

NHS
Antenatal and Newborn Screening Programmes



Homozygous a^+ Thalassaemia (Carrier

a - / a -



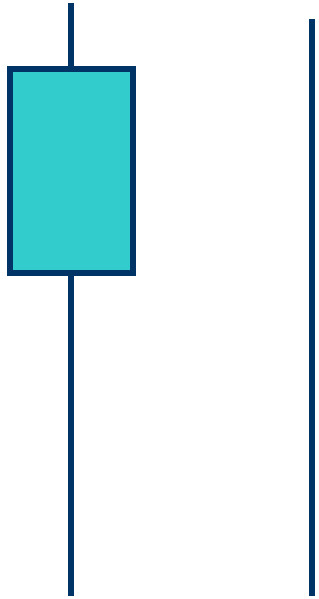
Heterozygous Thalassaemia (Carrier)

a a / - -

NSC
UK National Screening Committee

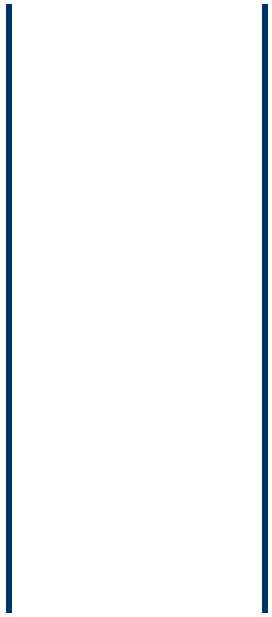
Alpha Thalassaemia

NHS
Antenatal and Newborn Screening Programmes



Haemoglobin H Disease

a - / - -



Haemoglobin Barts Hydrops Fetalis

- - / - -

Incompatible with Life

Beta Thalassaemia

- Full Blood Count
- The carrier state is often confused with iron deficiency due to reduced *MCV and MCH
- Hb A2 above 3.5 - diagnostic Normal 1.5 - 3.0 %
- DNA confirmation often required

ASYMPTOMATIC

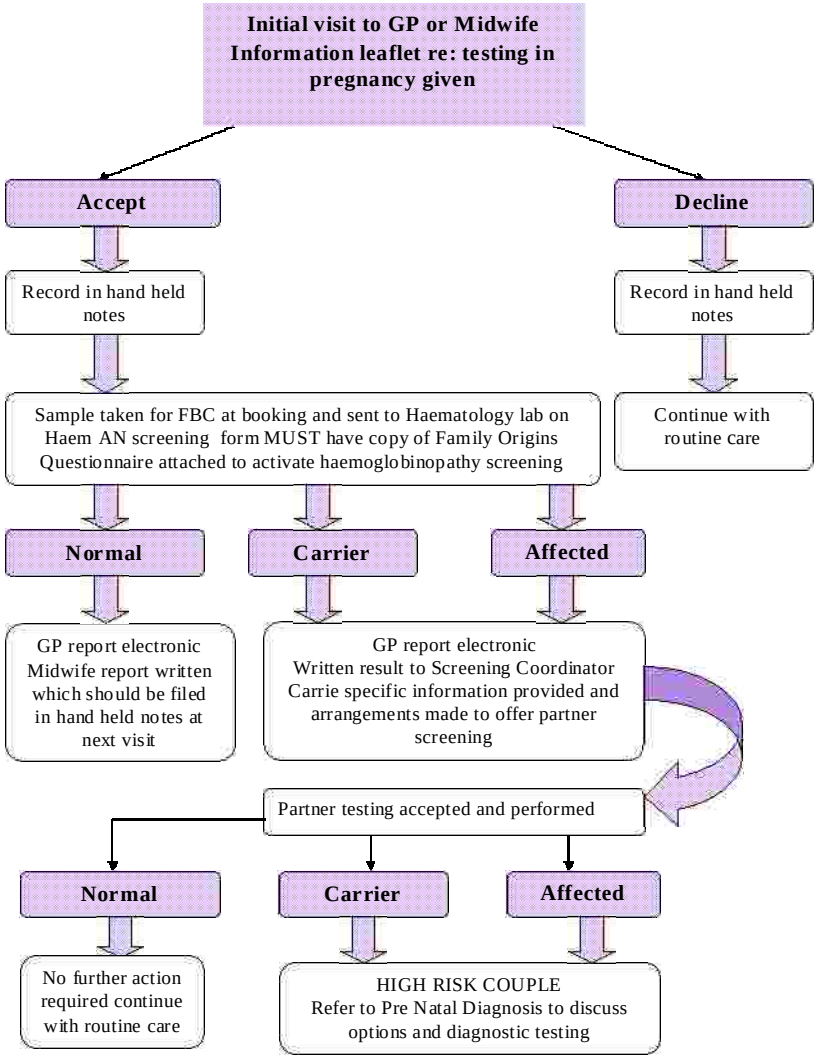
*Mean Corpuscular(Cell) Volume & Mean Corpuscular Haemoglobin

Carrier Frequency for Beta Thalassaemia

Affects up to:

- 1 in 7 Cypriots / Greek
- 1 in 10 Mediterranean
- 1 in 10 - 30 Asian / Middle East
- 1 in 30 Far East
- 1 in 25 - 100 African Caribbean
- 1 in 1000 Caucasian

Sickle Cell Disease and Thalassaemia Screening



Antenatal Screening last quarter

- 16 Confirmed carriers identified

- 87% of partners consented to screening

- q 11 high risk couples
  - q 2 Pre Natal Diagnosis
    - q 1 carrier- Delivered AS confirmed on neonatal testing
    - q 1 Sickle Cell Disease - TOP
  - q 9 Declined PND
    - q 5 non carriers
    - q 4 carriers

Newborn Blood Spot Test

NHS
Antenatal and Newborn Screening Programmes

NSC
UK National Screening Committee

What do we screen for?

- Phenylketonuria (PKU)
- Congenital Hypothyroidism (CHT)
- Sickle cell disorders (SCD)
- Cystic fibrosis (CF)
- MCADD from January 2009

What resources are available to support communication?

- National pre-screening

leaflet now in screening booklet and available in about 14 languages to download

- Results leaflets on CF,

CHT, PKU, SCD and MCADD are also available to download

Phenylketonuria (PKU)

- Affects 1 in 10,000 babies in UK (ie about 66 born each year)
- Inherited condition – carriers not identified
- Babies with condition are unable to digest phenylalanine (in protein)
- Untreated babies develop serious, irreversible, mental disability
- Early treatment with a strictly controlled diet prevents disability
- Treatment should start by 21 days of age

    - 2 borderline case in current year
    - 1further studies suggest NOT PKU
    - 1 baby diagnosed with galactosaemia

Congenital Hypothyroidism (CHT)

NHS Antenatal and Newborn Screening Programmes

- Affects 1 in 4,000 babies in UK (ie about 150 born each year)
- 1 in 10 cases are inherited – carriers not identified
- Babies with condition do not have enough thyroxine
- Untreated babies develop serious, permanent, physical and mental disability
- Early treatment with thyroxine tablets prevents disability
- Treatment should start by 21 days of age

    - 3 positive
    - 2 borderline

Sickle cell disorders (SCD)

- Affects 1 in 2,500 babies in UK (ie about 240 born each year)
- Inherited condition – carriers identified
- Red blood cells become sickle shaped
- Causes pain, tissue damage, infection and even death
- Early treatment through immunisations and antibiotics, as well as parent education, improves health and prevents deaths
- Treatment should be started by 2 months of age

    - 3 babies with Sickle cell non unexpected
    - 1 baby ? Sickle cell but left UK

Cystic fibrosis (CF)

NHS
Antenatal and Newborn Screening Programmes

- Affects 1 in 2,500 babies in UK (ie about 240 born each year)
- Inherited condition – carriers identified
- For some babies identified as carriers, CF cannot be ruled out
- Affects digestion and lungs, babies fail to thrive
- Screening avoids long delays in diagnosis
- Early treatment may improve health – cannot prevent the progression of the condition
- Treatment is with diet, medication and physiotherapy
    - 2 affected babies in Oxfordshire this year
        - 1 anticipated AN
        - 1 sibling parents chose not to be tested in pregnancy
    - 1 carrier in Oxfordshire

Medium Chain Aycl Co-A Dehydrogenase Deficiency

- Autosomal recessive
- Affects approx 1:10000 babies
- Affects the breakdown of fat and blocks energy production
- Leads to drowsiness, lethargy, vomiting, seizures and in some cases coma and death
- Symptoms can occur quickly in infants who are not feeding well or who have an intercurrent infection
- 20-28% mortality
- 30% of survivors have CNS sequelae at first clinical presentation
- Treatment is prevention of metabolic crisis
- Avoid fasting
- Early implementation of emergency regimes using Glucose polymers or IV dextrose

MCADD

- Identified by lab
- Screening coordinator and Paediatrician informed
- GP and HV contacted for information
- Family seen within 24 hours for 2nd line investigations and emergency regime
- Appointment with Metabolic specialist at GOS within 1 week

Any Questions?

THANK YOU